



Greig cephalopolysyndactyly syndrome

Greig cephalopolysyndactyly syndrome is a disorder that affects development of the limbs, head, and face. The features of this syndrome are highly variable, ranging from very mild to severe. People with this condition typically have one or more extra fingers or toes (polydactyly) or an abnormally wide thumb or big toe (hallux). The skin between the fingers and toes may be fused (cutaneous syndactyly). This disorder is also characterized by widely spaced eyes (ocular hypertelorism), an abnormally large head size (macrocephaly), and a high, prominent forehead. Rarely, affected individuals may have more serious medical problems including seizures, delayed development, and intellectual disability.

Frequency

This condition is very rare; its prevalence is unknown.

Genetic Changes

Mutations in the *GLI3* gene cause Greig cephalopolysyndactyly syndrome. This gene provides instructions for making a protein that controls gene expression, which is a process that regulates whether genes are turned on or off in particular cells. By interacting with certain genes at specific times during development, the *GLI3* protein plays a role in the normal shaping (patterning) of many organs and tissues before birth.

Different genetic changes involving the *GLI3* gene can cause Greig cephalopolysyndactyly syndrome. In some cases, the condition results from a chromosomal abnormality—such as a large deletion or rearrangement of genetic material—in the region of chromosome 7 that contains the *GLI3* gene. In other cases, a mutation in the *GLI3* gene itself is responsible for the disorder. Each of these genetic changes prevents one copy of the gene in each cell from producing any functional protein. It is unclear how a reduced amount of this protein disrupts early development and causes the characteristic features of Greig cephalopolysyndactyly syndrome.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one altered or missing copy of the *GLI3* gene in each cell is sufficient to cause the disorder. In some cases, an affected person inherits a gene mutation or chromosomal abnormality from one affected parent. Other cases occur in people with no history of the condition in their family.

Other Names for This Condition

- cephalopolysyndactyly syndrome
- GCPS

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Greig cephalopolysyndactyly syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265306/>

Other Diagnosis and Management Resources

- GeneReview: Greig Cephalopolysyndactyly Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1446>
- MedlinePlus Encyclopedia: Polydactyly
<https://medlineplus.gov/ency/article/003176.htm>
- MedlinePlus Encyclopedia: Syndactyly (image)
<https://medlineplus.gov/ency/imagepages/1763.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Polydactyly
<https://medlineplus.gov/ency/article/003176.htm>
- Encyclopedia: Syndactyly (image)
<https://medlineplus.gov/ency/imagepages/1763.htm>
- Health Topic: Craniofacial Abnormalities
<https://medlineplus.gov/craniofacialabnormalities.html>

- Health Topic: Hand Injuries and Disorders
<https://medlineplus.gov/handinjuriesanddisorders.html>
- Health Topic: Toe Injuries and Disorders
<https://medlineplus.gov/toeinjuriesanddisorders.html>

Genetic and Rare Diseases Information Center

- Greig cephalopolysyndactyly syndrome
<https://rarediseases.info.nih.gov/diseases/6550/greig-cephalopolysyndactyly-syndrome>

Educational Resources

- Disease InfoSearch: Greig Cephalopolysyndactyly Syndrome
<http://www.diseaseinfosearch.org/Greig+Cephalopolysyndactyly+Syndrome/3182>
- MalaCards: greig cephalopolysyndactyly syndrome
http://www.malacards.org/card/greig_cephalopolysyndactyly_syndrome
- Orphanet: Greig cephalopolysyndactyly syndrome
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=380

Patient Support and Advocacy Resources

- Children's Craniofacial Association
<http://www.ccakids.com/>
- myFace
<https://www.myface.org/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/greig-cephalopolysyndactyly-syndrome/>

GeneReviews

- Greig Cephalopolysyndactyly Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1446>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28greig+cephalopolysyndactyly+syndrome%5BTIAB%5D%29+OR+%28cephalopolysyndactyly+syndrome%5BTIAB%5D%29+OR+%28greig+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- GREIG CEPHALOPOLYSYNDACTYLY SYNDROME
<http://omim.org/entry/175700>

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